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Original Research Paper

Pediatrics

CASE REPORT OF INCIDENTAL DISCOVERY OF KIPPEL FEIL SYNDROME BEYOND THE EXPECTED CLINICAL TRIAD IN KVGMCH , SULLIA

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ABSTRACT

Introduction: Klippel–Feil syndrome (KFS) is a rare congenital cervical fusion condition, first described by Maurice Klippel and Andre Feil in 1912 . It is characterized by the congenital fusion of ≥2 cervical vertebrae, resulting from abnormal embryonic development during the first 3–8 weeks of gestation. The typical clinical triad of a low posterior hairline, a short neck, and restricted neck motion was originally recognized as the hallmark of KFS , but numerous spinal and extraspinal anomalies have now been documented in KFS patients The global incidence of KFS is estimated to be 0.71% , and the condition mainly affects female individuals (60% cases) **Case Report:** A 7 year old girl with a previous history of reduced hearing and small stature ,presented to KVG Pediatrics OPD with complaints of restricted neck movements, associated with pain since 2 weeks torticollis since 4 years of life on examination she had syndrome facies, short neck ,low posterior hairline , torticollis, scoliosis, SPRENGEL DEFORMITY, uneven shoulder ,sensorineural hearing loss for further evaluation x ray of cervical spine ,ultrasonography neck, was done computed tomography of thorax was done child was managed with following advise neck exercises, cervical braces and hearing aid with continue observation and monitoring., Further more to prevent spinal injuries, the patient was advised to avoid contact sports and activities associated with increased risk of head or neck movement. In case of development of any neurological symptoms, especially involving the upper extremities, the patient was advised to seek immediate medical attention **Conclusion-** There is no cure for Klippel-Feil syndrome (KFS), so treatments focus on managing the symptoms. Treatment programs can vary widely, depending on the severity of KFS, as well as any other conditions that might be present

KEYWORDS : Congenital, Fusion, Klippel-Feil Syndrome, Cervical, Vertebrae

INTRODUCTION

Klippel–Feil syndrome (KFS), known as cervical vertebral fusion syndrome, rare congenital condition characterized by abnormal fusion of any two of the seven bones in the neck (cervical vertebrae).[1]: It can result in a limited ability to move the neck and shortness of the neck, resulting in the appearance of a low hairline. Most people only have one or two of those symptoms so it may not be noticeable without medical imaging.[2]

Klippel-Feil syndrome Other names Congenital dystrophia brevicollis, cervical vertebral fusion syndrome.

Case Report

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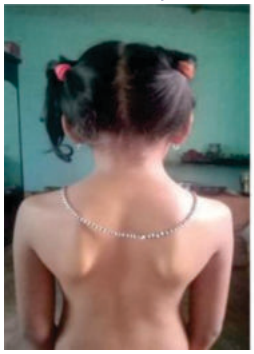
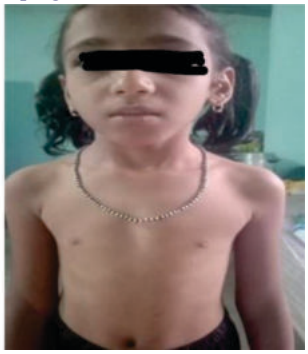
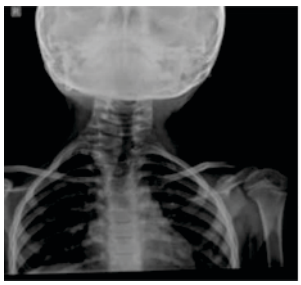


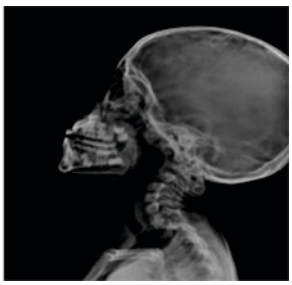


A 7 year old girl with a previous history of reduced hearing and small stature ,presented to kvg medical college pediatrics outpatient department with complaints of restricted neck movements ,associated with pain since 2 weeks torticollis since 4 years of life on examination Syndromic facies (triangular,broad forehead, wide and deep set eyes, pointed chin) Short webbed neck. Torticollis (head tilted to left side, low posterior hair line ,distance between ear pinna to trapezius upper border is grossly short compare to right side) SCOLIOSIS + left side of neck (cervico dorsal mild form) ,range of movement: full flexion, partly extension can. Be performed. Rotation and lateral bending showed limitation of movement. SPRENGEL deformity (Shoulder scapula is elevated on left side.) Slightly uneven shoulders sensorineural hearing loss on left side were noted (rinne test positive on left side, weber's test lateralized to right side/better.

Investigation

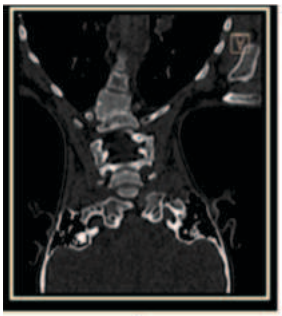
For further evalation x ray of cervical spine was done which showed –Bilateral cervical ribs noted with absence of left first rib ,mid cervical scoliosis to right ,widely spaced pedicles noted from c5 to t 1

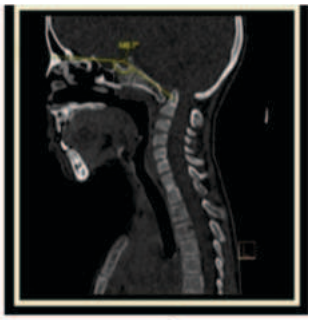


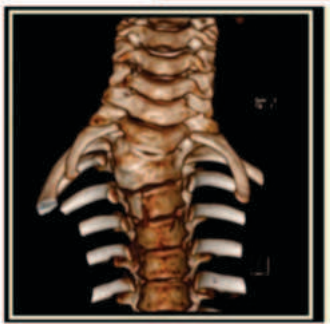


- Ultrasonography neck showed mild asymmetry of left sternocleidomastoid muscle , ultrasonography abdomen was normal
- Computed tomography thorax was done which showed –platybasia ,atlanto –occipital assimilation and basilar invagination seen ,Hypoplastic T 1 thoracic vertebral boby with fusion of T1 and T2 vertebrae ,Absent left T 1 hemivertebra and left 1st rib ,bilateral cervical ribs child was managed with following advise neck exercises, cervical braces and hearing aid with continued observation and monitoring., Further more to prevent spinal injuries, the patient was advised to avoid contact sports and activities associated with increased risk of

head or neck movement .In case of development of any neurological symptoms, especially involving the upper extremities, the patient was advised to seek immediate medical attention







DISCUSSION

ETIOLOGY - of Klippel-Feil syndrome is not well known [3]. Several studies have hypothesized that vascular disruption, global fetal insult, primary neural tube complications, or related genetic factors may carry implications in this condition's development [4][. Klippel-Feil syndrome may be present with fetal alcohol syndrome, Goldenhar syndrome, and Sprengel deformity [5[. Klippel-Feil syndrome can be caused by heritable mutations in the GDF6, GDF3, and MEOX1 genes. GDF6 and GDF3 influence embryonic bone development. The MEOX1 gene encodes the homeobox protein MOX1, which regulates vertebral separation.

GDF6 and GDF3 abnormalities are inherited in an autosomal dominant pattern, while MEOX1 mutations are autosomal recessive[6]

Epidemiology

klippel-Feil syndrome affects approximately 1 in 40,000 to 42,000 newborns worldwide, with a slight female predominance. A study by Nouri et al demonstrated a 2.0% incidence of Klippel-Feil syndrome on magnetic resonance imaging (MRI) in a global cohort of 458 patients [7]. In addition, Brown et al reviewed 1400 skeletons and reported a 0.71% incidence of the condition [8]. Notably, Klippel-Feil syndrome may also be asymptomatic. Children who do not undergo cervical imaging or present with an obvious physical deformity are likely to grow into adulthood unaware of their condition.[9]

Pathophysiology

Faulty cervical spine segmentation occurs during weeks 3 to 8 of embryonic development, resulting in persistent fusion of the involved vertebrae.[10] The Samartzis classification scheme is frequently used to describe Klippel-Feil syndrome fusions.[11] Type I involves a single congenitally fused segment, whereas type II involves multiple, noncontiguous, congenitally fused segments. Type III consists of multiple, contiguous, congenitally fused segments. About 25% of patients with Klippel-Feil syndrome present with type I deformities, while 50% have type II, and another 25% develop type III deformities.

CLASSIFICATION

- KFS classification

KFS classification

Type I	Extensive fusion of most or all of the cervical spine	Sporadic mutation
Type II	Fusion or only 1 or 2 vertebrae in the cervical spine	Autosomal dominant
Type III	Fusion exists in part of the thoracic and/or lumbar spine, in addition to Type I or Type II KFS	Autosomal recessive

- Despite this original classification, none is widely accepted
  - extent and locations of fusions is important
    - fusions above C3
      - more likely to be symptomatic and require abstaining from contact sports
    - fusions below C3
      - less likely to be symptomatic and most likely to have a normal life span
- Samartzis classification
  - carries prognostic value

Samartzis classification (2006)

Type I	Single-level congenital fusion of cervical segment	symptoms tend to be more axial
Type II	Multiple, noncontiguous congenitally fused segments	tend to develop peripheral myelopathy or radiculopathy
Type III	Multiple, contiguous congenitally fused segments in the cervical region	tend to develop peripheral myelopathy or radiculopathy

Differential Diagnosis

- Congenital scoliosis
- Postinfectious /spine inflammatory disorder
- Mayer – rokitansky –kaster syndrome
- Sprengel's deformity

TREATMENT

Most patients with Klippel-Feil syndrome receive nonoperative management, except in cases of acute neurological deficits, cervical instability, or chronic neurological issues that pose risks, necessitating operative management.[12]

The general treatment approaches to this condition are explained below.

Nonoperative Management Treatment for Klippel-Feil syndrome typically involves conservative measures tailored to manage symptoms. Patients with 1- or 2-level fusions below C3 may undergo monitoring and nonoperative management.

Patients can engage in contact sports such as hockey and rugby with appropriate education. However, individuals at higher risk of spinal deformity, particularly those with cervical fusion above C3, especially extending to the occiput, and long cervical spine fusions, should consider activity modification. Avoiding contact sports is crucial for these patients due to their increased risk of symptoms and susceptibility to spinal injuries. Clinicians should remember that many individuals with Klippel-Feil syndrome have polysyndromic presentations.

Operative Management

- Patients with persistent neurological pain, myelopathy, new-onset muscle group weakness, and documented spinal instability are operative candidates.[13]
- Surgical decision-making is influenced by spinal deformities and instability. Depending on clinical evaluation, surgeons may opt for cervical fusion using either an anterior or posterior approach. The anterior approach involves procedures such as anterior cervical fusion or corpectomy with synthetic or bone graft placement.[14]
- Cervical total disc arthroplasty is being investigated as a surgical option, showing promise in enhancing quality of life and preventing adjacent-level disease in adults with degenerative conditions.
- Posterior approaches, such as decompression and fusion, are viable options for treatment. In severe deformities, a combined anterior-posterior approach may be considered
- Additionally, surgical or bracing intervention may be necessary for associated compensatory thoracic scoliosis.

Prognosis

Patients with Klippel-Feil syndrome and cervical fusion above C3 tend to be more symptomatic. Research by Samartzis et al indicates that approximately two-thirds of individuals with this condition remain asymptomatic for over 8 years. Among those affected, individuals with type I deformity tend to experience more axial symptoms, while those with types II and III may develop myelopathy and radiculopathy

Complications

Congenital cervical fusion may predispose individuals to various conditions, necessitating monitoring and prompt management. These conditions include fractures, adjacent segment disease, disc degeneration, spondylosis, spinal canal stenosis, disc herniation, and osteophyte formation. Timely intervention, including surgical management when necessary, can mitigate complications and improve outcomes for individuals with Klippel-Feil syndrome.

Written Informed Consent Was Obtained From The Patient's Father For Publication Of This Case Report

CONCLUSION

- A rare case of Klippel Feil Syndrome is being presented with the aim that such cases should be identified and treated at an early stage to minimize cosmetic & social stigma to her and to her parents.
- The prevalence of patients with Klippel–Feil syndrome requiring surgery was 18.5%, with the majority undergoing posterior cervical surgery. Gender and instability were indentified as independent risk factors leading to surgical treatment

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